## Sandhoff Disease

Sandhoff disease is a rare, genetic, lipid storage disorder resulting in the progressive deterioration of the central nervous system. It is caused by a deficiency of the enzyme hexosaminidase which results in the accumulation of certain fats (lipids) in the brain and other organs of the body. Although Sandhoff disease is a severe form of Tay-Sachs disease--which is prevalent only in people of European Jewish descent--it is not limited to any ethnic group. Onset of the disorder usually occurs at 6 months of age. Symptoms may include motor weakness, startle reaction to sound, early blindness, progressive mental and motor deterioration, frequent respiratory infections, macrocephaly (an abnormally enlarged head), doll-like facial appearance, cherry-red spots, seizures, and myoclonus (shock-like contractions of a muscle).

There is no specific treatment for Sandhoff disease. Treatment is symptomatic and supportive.

The prognosis for individuals with Sandhoff disease is poor. Death usually occurs by age 3 and is generally caused by respiratory infections.

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